Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCHAD)

A fatty acid oxidation disorder

What is it?

Very Long Chain Acyl-CoA Dehydrogenase Deficiency (also known as VLCHAD) is an inherited fatty acid oxidation disorder. Patients with fatty acid oxidation disorders, like VLCHAD, cannot properly breakdown fats to energy. Once the body uses up its primary source of energy (glucose, or blood sugar), the body begins to fail because it cannot then make energy from fats. Therefore, people with VLCHAD must eat on a very regular basis and should not go long without food.

What are the symptoms?

A person with VLCHAD can appear normal at birth. Symptoms are variable and range from recurrent episodes of hypoglycemia, to heart and liver problems. These symptoms can progress very quickly to coma, cardiac arrest, brain damage, or even death in children who are not eating well. Many symptoms of VLCHAD can be prevented by immediate treatment and lifelong management. People with VLCHAD typically receive follow-up care by a team of professionals that is experienced in treating people with metabolic disorders.

Inheritance and frequency

VLCHAD is inherited in an autosomal recessive manner. This means that for a person to be affected with VLCHAD, he or she must have inherited two non-working copies of the gene responsible for causing VLCHAD. Usually, both parents of a person affected with an autosomal recessive disorder are unaffected because they are carriers. This means that they have one working copy of the gene, and one non-working copy of the gene. When both parents are carriers, there is a 1 in 4 (or 25%) chance that both parents will pass on the non working copies of their gene, causing the baby to have VLCHAD. Typically, there is no family history of VLCHAD in an affected person. VLCHAD is a rare fatty acid oxidation disorder; the total number of people affected with VLCHAD is not known.

How is it detected?

VLCHAD may be detected through newborn screening. A recognizable pattern of elevated chemicals alerts the laboratory that a baby may be affected. Confirmation of newborn screening results is required to make a firm diagnosis. This is usually done by a physician that specializes in metabolic conditions, or a primary care physician.

How is it treated?

VLCHAD is treated by eating frequently and avoiding fasting, and sometimes special medication, as recommended by a genetic metabolic medical specialist.

DISCLAIMER: This information is not intended to replace the advice of a genetic metabolic medical professional.

For more information:

Genetics Home Reference
Website: http://www.ghr.nlm.nih.gov

Save Babies Through Screening Foundation
4 Manor View Circle
Malvern, PA 19355-1622
Toll Free Phone: 1-888-454-3383
Fax: (610) 993-0545
Email: email@savebabies.org
Website: http://www.savebabies.org

FOD (Fatty Oxidation Disorder) Family Support Group
1559 New Garden Rd, 2E
Greensboro, NC 27410
Phone: (336) 547-8682 [8am - 8pm EST every day]
Fax: (336) 292-0536 [email/call ahead between 8am and 8pm before faxing]
Email: deb@fodsupport.org
Website: http://www.fodsupport.org

United Mitochondrial Disease Foundation
8085 Saltsburg Road, Suite 201
Pittsburgh, PA 15239
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